Letter to the Editor

Hydrometrocolpos as a Neonatal Manifestation of the Bardet-Biedl Syndrome

To the Editor:

We read with great interest the article "Genital Abnormalities in Females with Bardet-Biedl Syndrome" by Stoler et al. [1995]. In fact, we have observed 2 sisters with the Bardet-Biedl syndrome, one of whom manifested congenital hydrometrocolpos, a structural genital malformation which is rare in this syndrome. We agree with the authors of the paper cited above and re-emphasize that in a child with genital anomalies such as vaginal atresia and other appropriate manifestations the diagnosis of Bardet-Biedl syndrome should come to mind.

Sib 1 is a black girl who was born at term with a distended abdomen, imperforate vagina, and tetramelic postaxial polydactyly. A retrograde cystogram documented a dilatation of the uterus and a fistula between the bladder and the uterus. Patient underwent exploratory laparotomy and creation of a vaginal canal. Fluid aspiration confirmed the diagnosis of hydrometrocolpos. Intravenous pyelogram showed some blunting of the pelvocalyceal systems with bilateral hydronephrotic changes which were attributed to post-obstructive changes from the patient's hydrometrocolpos. Voiding cystourethrogram demonstrated no vesicoureteral reflux.

She presented to the eye clinic as a teenager with complaints of poor vision. At that time, she had mental deficiency, obesity, poor visual acuity, end gaze nystagmus, alternating exotropia, tapetoretinal degeneration, and extinguished electroretinogram. Speech was immature, rapid alternating movements were impaired, deep tendon reflexes were symmetrical, and she had difficulty with tandem gait. Cognitively, she was able to copy figures at the six-year level and do simple mathematics. She demonstrated dysmetria on finger-to-nose testing and dysdiadodokinesia. In addition, she had developed recurrent urinary tract infections. Repeat intravenous pyelogram showed bilateral caliceal clubbing and focal parenchymal loss. Voiding cystourethrogram was normal. Renal function was adequate with a blood urea nitrogen of 22 and a serum creatinine of 1.4. Menarche had begun at 14 years of age but was still irregular. The diagnosis of Bardet-Biedl syndrome was made.

Sib 2 is the older sister of patient 1. She presented to the eye clinic with her sister for similar eye complaints. Her perinatal history had been unremarkable. She had been born with tetramelic postaxial polydactyly which had been corrected surgically. She had been healthy most of her life, although mental deficiency had also been recognized like her sister. Physical exam showed obesity, poor visual acuity, tapetoretinal degeneration, extinguished electroretinogram, severely impaired rapid-alternating movements, and some difficulty with tandem gait. Cognitively, she was able only to copy figures from the five-year level. Menarche was at age 14 and menses were regular. The diagnosis of Bardet-Biedl Syndrome was established based on the tapetoretinal degeneration, mental retardation, obesity, and polydactyly and was supported by the diagnosis in the sister. Family history of these sisters was unremarkable.

REFERENCE

Stoler JM, Herrin JT, Holmes LB (1995): Genital abnormalities in females with Bardet-Biedl syndrome. Am J Med Genet 55:276–278.

Naveen Mehrotra
Susan Taub
Robert F. Covert*
Departments of Pediatrics and Ophthalmology
University of Chicago
Pritzker School of Medicine and
University of Chicago Hospitals
Chicago, Illinois

*Correspondence to: Robert F. Covert, M.D., Sections of Neonatology, Department of Pediatrics MC6060, Wyler Children's Hospital, University of Chicago, 5841 S. Maryland Avenue, Chicago, IL 60637.

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